Poster sessions will be in the Exhibit Hall in Regency Ballroom R on:
- Monday, February 10 from 4:30-6:30pm
- Tuesday, February 11 from 4:30-6:30pm
- Wednesday, February 12 from 4:30-6:30pm

Abstracts numbered from 1-164 are assigned to be presented on Monday, February 10, 2020 from 4:30-6:30 PM. Posters must be placed by 4:00 PM on Monday, and need to be removed at 6:30 PM when the Monday poster session ends.

Abstracts numbered from 165-328 are assigned to be presented on Tuesday, February 11, 2020 from 4:30-6:30 PM. Posters may be placed any time after 9:30 AM on Tuesday, but must be placed by 4:00 PM on Tuesday, and will need to be removed at 6:30 PM when the Tuesday poster session ends.

Abstracts numbered from 329-436 & all Late-Breaking Abstracts are assigned to be presented on Wednesday, February 12, 2020 from 4:30-6:30 PM. Posters may be placed any time after 9:30 AM on Wednesday, but must be placed by 4:00 PM on Wednesday, and will need to be removed at 6:30 PM when the Wednesday poster session ends.

Any poster numbers not listed will not be presented as the author is unable to attend the conference.

It is the policy of WORLD*Symposium* to publish all abstracts with the list of authors exactly as the abstract was submitted to WORLD*Symposium*. The first author of the submitted abstract will be listed as the presenting author on the Preliminary Program, Agenda, and Poster List.

Monday, February 10 – Poster Presentations

1	Ibane Abasolo	Extracellular vesicles increase the enzymatic activity of lysosomal proteins and improve the efficacy of enzyme replacement therapy in Fabry disease
2	Mary-Alice Abbott	Characteristics of Pompe disease patients with and without the c. 32 13T>G (IVS1) variant: data from the Pompe Registry
3	Jacqueline Adam	Understanding Fabry in Families Study - the availability of pedigree testing, genetic counselling and understanding of inheritance across Fabry International Network countries
4	Jacqueline Adam	Audit of a global rare disease clinical trial support service
5	Heather Adams	Cross-validation of the Vineland-III with independent assessments of cognition and adaptive skills in CLN3 disease
6	Faiza Adrees	A one tertiary centre experience - is compliance with migalastat an issue in Fabry patients?
7	Patricio Aguiar	Cell-mediated immunity in Fabry patients submitted to enzyme replacement therapy
8	Patricio Aguiar	Short term effects of migalastat in cardiac structure: A case report
9	Alia Ahmed	Development of mucopolysaccharidosis or MPS specific Infant Physical Symptom Score (IPSS)
11	Fahad Alharbi	Clinical utility of total concentration of globotriaosylsphingosine (Lyso-Gb3) and its analogues in diagnosing Fabry disease
12	Nadia Ali	Psychological health in adults with Morquio syndrome type A after six months of enzyme replacement therapy
13	José Álvarez González	Morquio syndrome type A treatment with non-viral vector
14	Hernan Amartino	Pseudoneuronopathic form of Hunter syndrome
15	Luise Ammer	Hematopoietic stem cell transplantation in a patient with mucolipidosis type II - A five-year follow-up
16	Marcio Andrade-Campos	TRAZELGA: preliminary results of the Spanish prospective, multi-center follow-up study and immune activation markers in adult Gaucher disease patients treated with eliglustat
17	Marcio Andrade-Campos	Impact of immunoparesis on Gaucher disease (GD): results from a network relationship analysis of data at diagnosis of the patients included in the Spanish registry of GD
18	Marcela Aquino	Early enzyme replacement therapy in Wolman disease: Challenges and outcomes

19	Carolina Aranda	Infantile-onset Pompe disease and CRIM negative status: Immunomodulation with intravenous immunoglobulin as an alternative for regular immune tolerance induction
21	Joao Augusto	The Fabry cardiovascular phenotype: Aortic stiffness in Fabry disease is linked to left ventricular hypertrophy and myocardial inflammation but not storage or fibrosis
22	Christiane Auray-Blais	Mass spectrometry approach allowing correlations between podocyturia and glycosphingolipids in Fabry disease patients
23	Stephanie Austin	Complicated cases and the need for individualized follow up plans for children diagnosed with Pompe disease via newborn screening
24	Stephanie Austin	Extended treatment with VAL-1221, a novel protein targeting cytoplasmic glycogen, in patients with late-onset Pompe disease
25	Mahima Avanti	Effects of enzyme replacement therapy on bone density in late onset Pompe disease
26	Olulade Ayodele	Clinical characteristics and health care resource utilization for patients with mucopolysaccharidosis II in the US: A retrospective chart review
27	Ahmed Ayuna	Cardiac rhythm abnormalities - an undervalued cardiovascular risk in adult patients with mucopolysaccharidoses (MPS) - one centre experience.
28	Laurie Bailey	The importance of the autopsy in understanding rare disease: Defining the role of the genetics team
30	Anneliese Barth	Early enzyme replacement therapy in a CRIM positive classic infantile Pompe patient: 11-year follow-up of a still progressive disease
32	Nicholas Bascou	Neurodevelopmental outcomes of hematopoietic stem cell transplantation for mucopolysaccharidosis type II: A prospective, longitudinal study
33	Julie Batista	Characterization of anti-agalsidase beta antibody formation from clinical trials and the Fabry Registry
35	Michal Becker- Cohen	Parkinson prodromal features in a large cohort of Gaucher carriers
36	Michal Becker- Cohen	Safety and efficacy of rapid intravenous velaglucerase-alfa infusion in naïve patients with Gaucher disease
37	Christian Beetz	Plasma adiponectin is a potential biomarker for organ involvement in male Fabry disease patients
39	Soumeya Bekri	Parsing the phenotypic landscape of lysosomal diseases using integrative network analysis
41	Nadia Belmatoug	Transition in health care from childhood to adulthood for lysosomal diseases patients in France, current state and priorities: The TENALYS study
42	Angela Beltrame	Progressive hearing loss in the α-galactosidase A deficient rat model of Fabry disease
44	Donna Bernstein	Lysosomal acid lipase deficiency and hematologic cancer predisposition
45	Venkata Boddupalli	Delineating the role of myeloid cells and brain microglia in Gaucher disease
46	Shaun Bolton	International Niemann-Pick Disease Registry: The characteristics of ASMD and NPC patients
47	Madeleine Bordley	Long-term clinical outcomes of patients with mucopolysaccharidosis type II: A case series
48	Marcella Borges	Lysosomal acid lipase deficiency across ages: Unraveling the clinical spectrum of an under-recognized genetic disorder
49	Poulomee Bose	Early synaptic dysfunction in MPS IIIC
50	Rebecca Bower	Sleep disturbance in children with mucopolysaccharidosis (MPS) types II and III; a review of medication usage
51	Colm Bradley	Improvement program in evaluation and management of Gaucher disease
52	Alissa Brandes	Gene therapy PR006 increased progranulin levels and improved lysosomal related phenotypes in model systems
53	Elizabeth Braunlin	Cardiopulmonary assessment of adults and adolescents with MPS disorders
54	Joanna Brokowska	Coenzyme Q10 in Sanfilippo disease

55	Alexander Broomfield	Long-term ambulatory outcomes in MPS I (Hurler syndrome) patients after HSCT
56	Alexander Broomfield	Pulmonary function in paediatrically diagnosed MPS I
57	Jillian Brown	Intracerebroventricular sulfamidase delivery to the brain
58	Alberto Burlina	Newborn screening for MPS I: The clinical benefit
59	Alberto Burlina	High incidence of Gaucher disease in northeast Italy: Results from lysosomal newborn screening
60	Barbara Burton	Long-term treatment with elosulfase alfa has an acceptable safety profile for patients with Morquio syndrome type A: Real-world results from the Morquio A Registry Study (MARS)
61	Umut Cagin	Liver expression of secretable GAA rescues advanced Pompe disease at the biochemical, functional, and transcriptional level in Gaa-/- mice
62	Alejandra Camacho-Molina	Fabry disease, differential diagnosis or coexistence with multiple sclerosis? A new mutation identified
63	Jessica Cardenas	Development of a conceptual model for variant late-infantile neuronal ceroid lipofuscinoses type 7 (CLN7)
64	Daniela Castillo-García	Gaucheromas: A complication in 2 children despite enzyme replacement therapy
65	Magdalena Cerón-Rodriguez	Are genotype and biomarkers enough to initiate enzyme replacement therapy in Fabry disease? The value of the biopsy
66	Sheng-Kai Chang	To develop a fusion protein combined α-galacosidase A and insulin-like factor 2 for treatment of Fabry disease
67	Huma Cheema	Mutation spectrum of glucocerebrosidase gene in Pakistani patients with Gaucher disease
68	Huma Cheema	Phenotypic diversity of Niemann-Pick disease type C in Pakistani children
69	Pin-Wen Chen	The lysosphingolipids analysis of sphingolipidoses in high-risk screening
70	Xin Chen	Preclinical efficacy and safety evaluation of scAAV9/CLN7 gene replacement therapy in rodents
71	Yun-Ru Chen	Development of a new pharmacological chaperone therapeutic strategy for Fabry disease
72	Chihya Cheng	Asian hotspot Fabry mutation, IVS4+919G>A, evidence for founder effect and originated in Asia more than 800 years ago
73	Yin-Hsiu Chien	Newborn screening for Morquio syndrome: Results from the 8-plex assay for 70,000 newborns
74	Chloe Christensen	Enhancing cell culture conditions for MPS IIIB induced pluripotent stem cells: The impact of dysregulated heparan sulfate turnover on FGF2 signalling
75	Heather Church	Validation of ELISA and cellular uptake inhibition assays to detect and quantify antibodies raised against sebelipase alfa in lysosomal acid lipase deficiency
76	Bruno Coghi	What lies beneath: Unraveling Niemann-Pick disease type C in adults
77	Tanya Collin-Histed	Regional manager program of the International Gaucher Alliance: No patient left behind
78	Tanya Collin-Histed	A global disease patient registry for neuronopathic Gaucher
79	Kristina Cotter	Development of a real-world evidence platform for MPS III
80	Claudia Cozma	Glucosylsphingosine for the screening, diagnosis, monitoring and prediction in Gaucher disease
81	Claudia Cozma	Hyaline fibromatosis syndrome: Genetic, clinical and biochemical characterization of a large cohort of patients
82	Claudia Cozma	2-tier screening approach for the identification of hereditary angioedema type 1 & 2 patients
83	Lisa Crawford	Integrative discovery approach to identification of biomarkers in Fabry disease patient biofluids

84	Pablo Crujeiras	LINCE project: A fast diagnosis of CLN2 disease
85	Carmen Silvia Curiati Mendes	Atypical neuronal ceroid lipofuscinosis type 2 (CLN2 disease): A case report
86	Carmen Silvia Curiati Mendes	Brain MRI findings in patients with mucopolysaccharidosis type VI (MPS VI)
87	Simona D'Amore	Glucosylsphingosine as a biomarker of disease burden in Gaucher disease
88	Julia Dao	Cellular and immune response to migalastat therapy in Fabry disease patients
89	Pronabesh DasMahapatra	Agalsidase beta stabilizes cardiac outcomes in Fabry patients: A systematic literature review
90	Laura Davids	Health care practitioners' experience-based opinions on providing care for patients with a positive newborn screen for Pompe disease and mucopolysaccharidosis type I
91	Crystal Davis	Natural history of GM1 gangliosidosis mouse models generated by The Jackson Laboratory Rare and Orphan Disease Center
92	Emily de los Reyes	Single-dose AAV9-CLN6 gene transfer stabilizes motor and language function in CLN6-type Batten disease: interim results from the first clinical gene therapy trial
93	Francisco del Castillo	Efficiency of NGS-based gene panels as first-line screening tests for the diagnosis of lysosomal diseases
94	Mireia del Toro	Hydrocephalus in patients with MPS: A not so unusual complication
96	Bianca Dias	Pulmonary function and functional capacity in patients with mucopolysaccharidosis
97	George Diaz	Preliminary data from first clinical trial of enzyme replacement therapy with olipudase alfa in pediatric patients with chronic visceral and neurovisceral acid sphingomyelinase deficiency
98	Mazen Dimachkie	NEO1 and NEO-EXT studies: Long-term safety and exploratory efficacy of repeat avalglucosidase alfa dosing for 5.5 years in late-onset Pompe disease patients
99	Tama Dinur	Gaucher disease specific patients reported outcome measures: A mobile phone survey
100	Jenny Do	Longitudinal behavioral characterization of Gaucher-associated Parkinson murine models
101	Marissa Donovan	Preclinical development of SIG-005 for treatment of MPS I
102	Gabriela Dostalova	A rare bird of Czechia: Case of homozygous female Fabry disease patient
103	Ivan Doykov	Improved specificity for detection of Niemann-Pick disease type C and other glycosphingolipidoses using a multiplex bloodspot assay
104	Halil Dundar	Triamterene-induced suppression of R227X premature termination codon in Fabry disease
105	Mariola Edelmann	Exosomes with unique CNS-targeting properties brings novel therapeutic strategy to neuronopathic LDs
106	Grigorios Effraimidis	Globotriaosylsphingosine (Lyso-Gb3) and analogues in plasma and urine of Fabry disease patients and relation to long-term treatment in a nationwide female Danish cohort: A retrospective study of a prospective cohort
107	Grigorios Effraimidis	Cascade screening outcome in a nationwide cohort and genotype-phenotype relationship
108	Khaled Eid	Delineating D409H (D448H) homozygous phenotype-genotype in an international cohort of the International Collaborative Gaucher Group Gaucher Registry: Cardiac involvement and early mortality
109	Julie Eisengart	Evidence of problems with "processing efficiency" in attenuated mucopolysaccharidosis type I
110	Farah El Turk	Comparative and correlation study of biochemical substances in serum and urine of LDs patients
111	Fatma Eminoglu	Gaucher disease type 3: Variability in phenotype among siblings with same mutation
112	Takumi Era	Presynaptic dysfunction in neurons derived from Tay-Sachs-iPSCs

113	María José Esteban Giner	Enzymatic replacement therapy in Hunter syndrome (MPS II): A systematic review with narrative synthesis and meta-analysis
114	Francois Eyskens	The Belgian hematology project: Screening for Gaucher and Niemann-Pick disease
115	Qi Fan	Agalsidase beta slows the progression of renal outcomes in Fabry patients: A systematic literature review
116	Luca Fierro	Newborn screening for Pompe disease in New York state: Results from 6 year single center experience
117	Niamh Finnegan	Promoting independence and empowering patients on enzyme replacement therapy
118	Rachel Fisher	Prenatal diagnosis of non-infantile Sandhoff disease
119	Rachel Fisher	Second tier testing for newborn screening: The Michigan experience
121	Brian Fluharty	Preclinical development of SIG-007 for treatment of Fabry disease
122	Gandhy Fonseca-Gonzalez	Association of Fabry disease and cancer: Patient report and literature review
123	Stuart Forshaw-Hulme	Role of a specialist nurse in starting substrate reduction therapy in Gaucher disease type 1: One centre experience
124	Stuart Forshaw-Hulme	Understanding the biopsychosocial factors contributing to mental health issues in Fabry disease-one tertiary centre experience
125	Nicholas France	Sphingosine-1-phosphate receptor type 5 (S1P5) agonism: A potential new mechanism for the treatment of neuronopathic features of Niemann-Pick disease type C and neurodegenerative sphingolipidoses
126	Francesca Fumagalli	Lentiviral hematopoietic stem and progenitor cell gene therapy (HSPC-GT) for metachromatic leukodystrophy (MLD): Clinical outcomes from 33 patients
127	Chai Gadepalli	Nasendoscopy findings in adult patients with mucopolysaccharidosis- a tertiary UK centre experience
128	Lidia Gaffke	Changes in the vacuolar transport: Insight into pathomechanism of mucopolysaccharidosis
129	Marta Gandía	Identification of the missing causative mutations in monoallelic cases of late-onset Pompe disease (LOPD)
130	Eric Joshua Garcia	Predicting Parkinson disease: Statistical profiling of clinical data in an at-risk population
131	Michael Gelb	A universal newborn and diagnostic screening platform for lysosomal diseases and beyond
132	Michael Gelb	Glycosaminoglycan biomarkers in newborn dried blood spots to support newborn screening of MPS disorders
134	Jacinthe Gingras	AAVHSC characterization for developing treatments for human genetic diseases of the nervous system
136	Roberto Giugliani	Trends in age of diagnosis and time to treat for MPS I
137	Roberto Giugliani	Assessing the impact of the five senses on quality of life in mucopolysaccharidosis
138	Roberto Giugliani	Results from a phase 2 trial of a blood-brain barrier penetrating enzyme (JR-141) in patients with MPS II in Brazil
139	Ozlem Goker-Alpan	The correlation of Gaucher disease burden and downstream complement activation
140	Mehmet Goktas	Rare cause of protein losing enteropathy: Gaucher disease type 3
142	Esteban Gonzalez	Effect of losartan and propranolol on cardiac remodeling in mucopolysaccharidosis type I mice
143	Isabela Gonzalez	Follicular helper T-cell and B-cell subsets in mucopolysaccharidosis patients
144	Domingo González-Lamuño	Expression levels of the mono carboxylate transporter MCT1 in erythrocytes from patients with Fabry disease and predominant hypertrophic myocardiopathy
145	Domingo González-Lamuño	Vacuolated lymphocytes of blood smears in the diagnoses of lysosomal disorders
146	Antonio González-Meneses	Inflammatory mediators and cytokines in acid sphingomielinase deficiency (ASMD)
147	Antonio González-Meneses	Study of inflammatory mediators in different mucopolyssacharidosis patients.
148	Sharan Goobie	Intrafamilial variability in late-onset CLN2 disease

149	Christina Grant	Persistent EBV viremia in siblings with lysosomal acid lipase deficiency (LAL-D)
150	Christina Grant	Laronidase desensitization protocol in a fluid sensitive child after anaphylaxis during hematopoietic stem cell transplant
151	Elena Gras-Colomer	Population pharmacokinetic of enzymatic activity of glucocerebrosidase in Gaucher disease patients treated with enzyme replacement therapy
152	Samuel Gröschel	Effect of intrathecal recombinant human arylsulfatase A enzyme replacement therapy on structural brain MRI in children with metachromatic leukodystrophy
153	Amanda Gross	Cardiovascular manifestations of feline Sandhoff disease after intravenous AAV gene therapy
154	Punita Gupta	Homozygous TBCK mutation - A novel type of lysosomal disorder
155	William Hallows	Engineering α-galactosidase A (GLA) to improve protein stability, efficacy and reduced immune response for the treatment of Fabry disease
156	Sang-oh Han	Comparisons of infant and adult mice reveal age effects for liver depot gene therapy in Pompe disease
157	Paul Harmatz	A new randomized placebo controlled study to establish the safety and efficacy of velmanase alfa (human recombinant alpha-mannosidase) enzyme replacement therapy for the treatment of alpha-mannosidosis
158	Simon Heales	Inhibition of GBA2 may influence the neuronal response to oxidative stress: Implications for Gaucher and Parkinson disease
159	Laura Heckman	Gene therapy PR001 increased GCase activity and improved neuronopathic Gaucher disease phenotypes in mouse models
160	Coy Heldermon	Disease correction by intraparenchymal or cisternal delivery of a modified AAV8 capsid expressing codon optimized NAGLU for mucopolysaccharidosis type IIIB mice
161	Nadene Henderson	Alternative agalsidase beta dosing strategies in a combined cohort
162	Christian Hendriksz	Velmanase alfa enzyme replacement therapy for alpha-mannosidosis improves patient outcomes over standard of care both in terms of clinically relevant improvement and disease stabilization
163	Julia Hennermann	Puberty, fertility and pregnancy in patients with mucopolysaccharidosis and mucolipidosis: A multicentre cross-sectional study

Tuesday, February 11 – Poster Presentations

165	Alvaro Hermida	Arterial stiffness assessment in naïve patients with Fabry disease
166	Wendy Heywood	Development of a plasma lyso-Gb1 clinical assay and its application to Gaucher and Krabbe disease patient plasma
167	Hayriye Hizarcioglu-Gulsen	Biliary atresia and Niemann-Pick disease type C: Coincidence or a mimic?
168	Brittany Hodge	Patient-centered development of a pretest genetic counseling video for Fabry disease
169	Robert Hopkin	Improvement of Fabry disease-related gastrointestinal symptoms in significant proportions of classic male patients treated with agalsidase beta: A Fabry Registry analysis
170	Sinead Horgan	Chart review of the management of late-onset Pompe patients diagnosed through newborn screening
173	Caoimhe Howard	Adherence to international and local guidelines in Irish Morquio syndrome type A patients
175	Leroy Hubert	Molecular diagnostic findings of lysosomal diseases as a result of "Detect Lysosomal Storage Diseases", a no-charge sponsored testing program
176	Derralynn Hughes	Pegunigalsidase alfa, PEGylated α-galactosidase-A enzyme in development for the treatment of Fabry disease, shows correlation between renal GB3 inclusion clearance and reduction of plasma Lyso-GB3
177	Derralynn Hughes	A phase I/II multicenter gene therapy clinical study for Fabry disease
178	Derralynn Hughes	Prompt initiation of agalsidase alfa therapy is associated with improved cardiovascular and renal outcomes in the Fabry Outcome Survey (FOS)
179	Derralynn Hughes	First-in-human study of a liver-directed AAV gene therapy (FLT190) in Fabry disease
180	Jackie Imrie	The International Niemann-Pick Disease Registry (INPDR): A beacon for rare diseases
181	Linda Ingemann	Rescue of NPC1 protein and effect on biomarkers by arimoclomol treatment in Niemann-Pick disease type C
182	Kohji Itoh	Innovative gene therapy for lysosomal neuraminidase 1 (NEU1) deficiencies
183	Margarita Ivanova	Clinical spectrum and molecular variants in Gaucher disease presenting in early infancy
184	Ayuko Iverson	Sequencing-based screening for lysosomal disorders in a multi-ethnic biobank
185	Katharina Iwan	Deep phenotyping proteomics analysis of CSF from CLN2 patients undergoing enzyme replacement therapy
186	Emanuela Izzo	Utility of gene panel testing in children with seizure onset after 2 years of age: Results from a European and Middle Eastern epilepsy genetic testing program
187	Skyler Jackson	Methodologies that lead to conceptual strength and representation in rare disease qualitative research
188	Yaanu Jeyakumar	Neurodevelopmental outcomes in patients with mucopolysaccharidosis type 1 (MPS I)
189	Dipesalema Joel	The impact of providing rare disease educational programs in resource-limited settings
190	Franklin Johnson	Exposure-response of migalastat in support of extrapolation of efficacy from adults to children with Fabry disease
191	Ana Jovanovic	Airways abnormalities in adult patients with mucopolysaccharidosis, single institute experience
192	Aneta Kaczmarczyk	Analysis of heparan sulfate and heparan sulfate non-reducing ends in mucopolysaccharidosis type I
193	Shih-hsin Kan	Intra-articular AAV9 α-iduronidase gene therapy in mucopolysaccharidosis type I canine model
194	Adam Kanack	Platelet and myeloid cell phenotypes in a rat model of Fabry disease
195	Ilkka Kantola	PQ interval, QRS duration and QTc interval increased in Fabry patients treated by enzyme replacement therapy both for 10 and 15 years
196	Nesrin Karabul	First signs for lysosomal disorders (LD) - kidney ultrasound

197	George Karkashadze	Abnormalities in the cerebral cortex in Gaucher disease type 1: Findings from the ENIGMA storage disease working group
198	Reena Kartha	Synergistic chaperone activity of N-acetylcysteine and its metabolite L-cysteine in Gaucher disease
199	David Kasper	Challenges for newborn screening and rare disease diagnostic initiatives in Europe
200	David Kasper	Combined biochemical and targeted-next generation sequencing panel for differential diagnosis of inherited myopathies
201	David Kasper	The value of biochemical enzymatic testing for the rapid identification of early-onset Pompe disease in newborns and children
202	Asaka Katabuchi	GALC-folding assistant molecules as potential therapies for Krabbe disease
203	Asaka Katabuchi	Psychosine-reducing molecules as potential therapies for globoid-cell leukodystrophy or Krabbe disease
204	Marcel Kelkel	Analysis of urinary sphingolipids in patients with rare genetic diseases using a tandem mass spectrometry approach
205	Scott Kerns	Assessment of various routes of AAV administration in achieving CNS transduction
206	Scott Kerns	Combination AAV delivery to target vision loss and CNS manifestations in CLN3 disease
207	Brian Kevany	A novel AAV capsid with improved tropism to heart, kidney and PNS for treatment of Fabry disease
208	Brian Kevany	Intravenous delivery of a novel AAV capsid with improved PNS tropism reduces underlying Pompe disease pathology
209	Aleena Khan	Whole-body MRI in late-onset Pompe disease: Clinical utility and correlation with functional measures
210	Aleena Khan	Higher dosing of alglucosidase alfa improves outcomes in children with Pompe disease
211	IkHui Kho	Study of a novel neuraminidase 1 knockout mouse links the pathology of sialidosis in the nervous, renal and reproductive system
212	Katherine Kim	Development of high sustained IgG antibody titers and corresponding clinical decline in an adolescent with atypical infantile Pompe disease after 11+ years on enzyme replacement therapy with alglucosidase alfa
213	Sarah Kim	Quantification of cerebrospinal fluid chitotriosidase in a clinical laboratory is validated for use in diagnosis and clinical trials
214	Virginia Kimonis	A patient-reported outcome validation study of concept elicitation and cognitive debriefing to understand neuropathic pain in Fabry disease
215	Virginia Kimonis	Antisense oligonucleotide targeting glycogen synthase (GYS1) in a Pompe disease mouse model
217	Jennifer Klein	Using machine learning to identify chaperones for sialidosis
218	Dwight Koeberl	A phase 1 study of gene therapy with ACTUS-101 in late-onset Pompe disease
219	Youngil Koh	Germline variants of lysosomal disease has increased risk of cancer
221	Aditi Korlimarla	A new look at an old disease: Is Pompe disease a neuromuscular disorder with CNS involvement?
222	David Kronn	Mini-COMET study: Safety, immunogenicity, and preliminary efficacy for repeat avalglucosidase alfa dosing in patients with infantile-onset Pompe disease (IOPD) who were previously treated with alglucosidase alfa and demonstrated clinical decline
223	Francyne Kubaski	Quantitation of glycosaminoglycans in amniotic fluid by liquid chromatography tandem mass spectrometry: A potential tool for the rapid prenatal identification of MPS in pregnancies at risk
224	Francyne Kubaski	Quantification of glycosaminoglycans in mucopolysaccharidosis type IIID: The first Brazilian patient identified
225	Francyne Kubaski	Quantification of glycosaminoglycan species in patients with multiple sulfatase deficiency by liquid chromatography tandem mass spectrometry: A potential biomarker for this condition

226	Francyne Kubaski	Newborn screening for six lysosomal diseases: Pilot study in Brazil
227	Anja Lachmann	Home infusion therapy with agalsidase alfa for patients with Fabry disease in Germany and Austria
228	Dawn Laney	Longitudinal change in the urinary biomarkers of young pediatric patients with pathogenic variants in the GLA gene: Data from the MOPPet Study
229	Dawn Laney	Use of flotation-REST (restricted environmental stimulation technique) therapy in treatment of Fabry related pain
230	Heather Lau	Long-term efficacy and safety of vestronidase alfa enzyme replacement therapy in subjects with mucopolysaccharidosis type VII <5 years old
231	Lucia Lavalle	Relative contribution of acid ceramidase and alfa-galactosidase a to lyso-Gb3 and severity in Fabry disease
232	Maria Dolores Ledesma	Inhibition of fatty acid amide hydrolase prevents pathology in a mouse model of acid sphingomyelinase deficiency by rescuing downregulated endocannabinoid signalling
233	Chris Lee	Cell modeling and assay development for Krabbe disease
234	Katelyn Leestma	Ethical and psychosocial implications of mucopolysaccharidosis type I identified by newborn screening in the complex care setting
235	Kimmo Lehtimäki	Longitudinal characterization of the Cln8mnd-/- mouse model of CLN8 Batten disease fine motor performance, retinal degeneration, brain pathology, and metabolic changes
236	Malte Lenders	Fabry stabilization index (FASTEX): Clinical evaluation of disease progression in Fabry patients
237	Malte Lenders	Neutralizing anti-drug antibodies inhibit endothelial enzyme uptake and activity in Fabry disease
238	Renuka Limgala	Selective screening for lysosomal disorders in a large cohort of minority groups shows higher incidence rates and novel variants
239	Renuka Limgala	Assessing mast cell cross-talk with dendritic and natural killer cells for clinical management and efficacy of enzyme replacement therapy in Fabry disease
240	Ales Linhart	Switching from agalsidase alfa to pegunigalsidase alfa for treating Fabry disease: One year of treatment data from BRIDGE, a phase III open label study
241	Lin Liu	A new platform technology for next generation lysosomal enzyme replacement and potential gene therapy in the treatment of lysosomal diseases
242	Ying Poi Liu	Development of an AAV5-based gene therapy for Fabry disease
243	Benjamin Lohmöller	Successful ambroxol treatment in Gaucher disease type 2: Age-appropriate neurocognitive development after one year of follow-up
244	Valynne Long	A case report of successful treatment of teenage patient with lysosomal acid lipase deficiency
245	Mabel Lopez	A prospective natural history study of Krabbe disease in a patient cohort with onset between 0 and 12 months
246	Laura López de Frutos	Serum protein electrophoresis pattern alterations on lipidoses patients
247	Juan Losada	In silico identification of pharmacological chaperones for mucopolysaccharidosis type IIIB
248	Charles Lourenco	Infertility in Fabry disease: An overlooked feature in alfa-galactosidase deficiency?
249	Charles Lourenco	Late-onset Krabbe disease: Findings from a cohort of Brazilian patients
250	Sara Lucas-Del-Pozo	Amyloid deposition in a patient with the complex Gaucher disease/Parkinson disease
251	Nataniel Ludwig	Quantification of glycosaminoglycans by liquid chromatography tandem mass spectrometry is a useful tool for screening of GlcNAc-phosphotransferase deficient patients
252	Zoltan Lukacs	Comparison of tripeptidyl peptidase 1 (CLN2) measurement by fluorometry and tandem mass spectrometry
253	Zoltan Lukacs	Targeted-population screening for mucopolysaccharidoses: Results of the assessment of more than 9000 samples

254	Dinesh Lulla	Survey assessing the prevalence and severity of neuropsychiatric manifestations in patients living with Fabry disease
255	Troy Lund	New biomarkers for MPS IH: Plasma iduronidase and urine non-reducing ends
256	Erik Lykken	Combination intrathecal and intravenous gene therapy reveals a dominant role for treatment age in determining survival and behavioral outcomes in the mouse model of infantile neuronal ceroid lipofuscinosis
257	Nicole Lyn	Assessing upper extremity limitations of late-onset GM2 gangliosidoses with the Neuro-QOL Item Bank - Upper Extremity Function (Fine Motor, ADL) - Short Form and 9 Hole Peg Test
258	Ryuichi Mashima	Multiplex measurement of lysosomal disease enzyme activity using LC-MS/MS
259	Margaux Masten	Hamburg Late Infantile Neuronal Ceroid Lipofucinosis Scale (H-LINCLS) vs. the Unified Batten Disease Rating Scale (UBDRS): Comparison and cross-validation
260	Margaux Masten	CLN1 disease natural history data: Prospective and retrospective analysis
261	Margaux Masten	Age-at-onset of core features of CLN3 disease: A cross-sectional and longitudinal natural history study
262	Margaux Masten	The CLN3 Disease Staging System (CLN3SS): A tool for stratification based on disease severity
263	Fulvio Mavilio	Pre-clinical safety and efficacy findings of AT845, a novel gene replacement therapy for Pompe disease targeting skeletal muscle and heart
264	Maria Mazurkiewicz-Beldinska	Strategies to shorten diagnostic delays for late infantile neuronal ceroid lipofuscinosis type 2 (CLN2 disease)
266	Margaret McGovern	Prospective study of the natural history of chronic acid sphingomyelinase deficiency in children and adults: Eleven years of observation
267	R. Scott McIvor	Iduronidase-transposed human B lymphocytes correct enzyme deficiency and glycosaminoglycan storage disease in immunodeficient mucopolysaccharidosis type I mice
268	Thomas Mechtler	Analytical and diagnostic performance of a DBS based assay for GM1 and GM2
269	Thomas Mechtler	Lyso-sphingomyelin as biomarker for Niemann-Pick disease type A and B patients
270	Thomas Mechtler	Investigating the suitability of high-resolution mass spectrometry for newborn screening: Identification of hemoglobinopathies and β-thalassemia in dried blood spots
271	Lacie Mehr	Facial phenotyping in Fabry using Face2Gene
272	Eugen Mengel	Home infusion therapy with velaglucerase alfa for Gaucher disease type 1 in Germany and Austria
273	Jonathan Mink	Cross-sectional and longitudinal quantification of CLN3 disease progression
274	Carlos Miranda	One-off liver directed AAV gene therapy achieves long term uptake of acid beta-glucocerebrosidase by macrophages of affected tissues in Gaucher disease
275	Pramod Mistry	Individual patient responses to eliglustat in treatment-naïve adults with Gaucher disease type 1: Final data from the phase 3 ENGAGE trial
276	John Mitchell	Expanding the phenotype: Acid ceramidase deficiency presenting with features of SMA-PME and Farber disease
277	John Mitchell	Farber disease (acid ceramidase deficiency) natural history study: Prospective and retrospective clinical data
278	John Mitchell	Long-term clinical outcomes of patients treated with elosulfase alfa: Five-year real-world results from the Morquio A Registry Study (MARS)
279	Feda Mohamed	The pharmacological chaperone N-n-butyl-deoxygalactonojirimycin enhances beta-galactosidase processing and activity in an infantile GM1-gangliosidosis patient fibroblast cells
280	James Moon	Chronic myocardial edema in Fabry disease is linked to myocardial injury and left ventricular volume/pressure overload

281	Lina Mora	Bone impact of a late diagnosis in Gaucher disease
282	Marta Morado	Pompe disease: PAS-positive lymphocyte vacuoles as diagnostic screening test
283	David Moreno-Martínez	Stroke characterisation and description of identified risk factors in a Fabry disease large cohort
284	David Moreno-Martinez	Chronic immune thrombocytopenia refractory to treatment in a patient with Gaucher disease with a common genotype
285	Swati Mukherjee	Long-term clinical outcomes of patients with Morquio syndrome type A treated with elosulfase alfa: Results from a Managed Access Agreement in England
286	Behzad Najafian	Podocyte globotriaosylceramide (GL-3) content in female adult patients with Fabry disease and amenable mutations reduces following 6 months of treatment with migalastat
287	Juana Navarrete	Use of biomarkers to follow up positive lysosomal diseases in newborn screening
288	Igor Nestrasil	Intraspinal space restriction at the occipito-cervical junction alters cervical spinal cord diffusion MRI metrics in mucopolysacharidoses patients
289	Michelle Ng	Impact of extended post-HSCT enzyme replacement therapy (ERT) on linear growth in mucopolysaccharidosis type IH (MPS IH)
290	Miriam Nickel	Current understanding of language development in late infantile neuronal ceroid lipofuscinosis type 2 (CLN2 disease)
291	Elena-Raluca Nicoli	Targeting GLB1 in mice by CRISPR/Cas9 genome editing: Establishing a novel model for type II GM1 gangliosidosis
292	Vera Niederkofler	Neuroinflammation in mouse models of two different lysosomal diseases
293	Graeme Nimmo	Parkinsonism in a Canadian population of patients with type 1 Gaucher disease
295	Dau-Ming Niu	Development of a gene therapy for Fabry disease using adeno-associated viral vector mediated gene transfer
296	Albina Nowak	Circular RNAs expression profiles and their phenotype associations in Fabry disease: A pilot study
297	Antonio Ochoa-Ferraro	Audit on cardiovascular risk in Fabry disease
298	Loreanne Oh	Validation of an expert system-generated checklist for the early diagnosis of mucopolysaccharidosis type III A and B
299	Juho Oksman	Principal component analysis (PCA) based data fusion approach for a mouse model of CLN8 Batten disease.
300	Torayuki Okuyama	Therapy for mucopolysaccharidosis II with an intravenous blood-brain barrier-crossing enzyme (JR-141): 26-week results from a phase 3 study in Japan suggesting significant efficacy against central nervous system and systemic symptoms
301	Torayuki Okuyama	Successful prevention and stabilization of cognitive decline in Japanese patients with neuronopathic mucopolysaccharidosis type II treated by intracerebroventricular enzyme replacement therapy: Results of the Phase I/II clinical trial for two years.
302	Andrew Oldham	Long-term outcomes in adults affected with mucolipidosis type III: A two-centre experience.
303	Petra Oliva	Diagnostic strategy for females suspected of Fabry disease
304	Petra Oliva	Differential diagnosis for mucopolysaccharidoses (MPSs)
305	Petra Oliva	Differential diagnosis of Niemann-Pick disease types A and B in cases of suspected Gaucher disease
306	Cara O'Neill	Parent prioritization of meaningful treatment targets for Sanfilippo syndrome
307	Cara O'Neill	PROVIDE: Video-based patient-reported outcomes for Sanfilippo syndrome: A new and innovative approach to record and measure disease post-gene therapy
308	Neslihan Önenli Mungan	First case report of Gaucher disease and Graves' thyroiditis
311	Chris Orsborne	Migalastat therapy for Fabry disease; real-word clinical outcomes comparing those previously treated with ERT with those who are treatment naïve

312	Chris Orsborne	A retrospective outcome analysis of chaperone therapy in Fabry disease: Clinical outcomes after the first year of therapy - a single centre experience
313	Chris Orsborne	The clinical features of advanced Fabry disease: A single centre, twelve-year experience.
314	Saida Ortolano	Effect of enzyme replacement therapy on the innate and adaptative immune response in patients with Fabry disease: Preliminary results
315	Liliya Osipova	Frequency of epilepsy in neuronopathic mucopolysaccharidoses
316	Li Ou	Liver-targeting gene editing achieves significant neurological benefits in MPS I mice
317	Li Ou	Correction of both Tay-Sachs and Sandhoff diseases with the PS gene editing system
318	Harry Pachajoa	Disability experience of a women with Morquio syndrome type IVA
320	Francis Pang	Caregiver-reported impact on quality of life and disease burden in patients diagnosed with metachromatic leukodystrophy: Results of an online survey and a qualitative interview
321	Tiffany Pang	Update on a no-cost epilepsy gene panel for seizure onset between 2-4 years of age: Results from 682 tests
322	Marc Patterson	Efficacy and safety of arimoclomol in patients with Niemann-Pick disease type C: Results from a double-blind, randomized placebo-controlled trial with a novel treatment
323	Loren Pena	Impact of early diagnosis by NBS on existing genotype phenotype correlations for Pompe disease
324	Merlene Peter	An overview of the reproductive system in mucopolysaccharidosis and case control study to identify menstrual and pregnancy complications in women with MPS
325	M. Judith Peterschmitt	Safety, pharmacokinetics, and pharmacodynamics of oral venglustat in Parkinson disease patients with a GBA mutation from Japan and the rest of the world: Results from part 1 of the MOVES-PD study
326	Dawn Phillips	Maroteaux-Lamy syndrome (mucopolysaccharidosis type VI): Symptoms and the impact on function and activities of daily living (ADL)
327	Karolina Pierzynowska	Disturbancy in the efficiency of macromolecule degradation systems as one of the unknown aspects of the pathogenesis of mucopolysaccharidoses
328	Stacy Pike-Langenfeld	Disease burden and treatment considerations in Krabbe disease: The caregiver perspective

Wednesday, February 12 – Poster Presentations

329	Chelsie Poffenberger	LRRK2 findings in a family with GBA1 mutations: A case study and implications for genetic counseling
330	Lynda Polgreen	Growth, bone, and joint outcomes in non-neuronopathic mucopolysaccharidosis type II over 10 years
331	Juan Politei	Fabry disease patient-reported outcome-gastrointestinal (FABPRO-GI): A new Fabry disease-specific gastrointestinal outcomes instrument
332	Laura Pollard	High prevalence of pseudodeficiency for MPS I and MPS II and the impact on newborn screening
334	Michael Przybilla	Gene editing to treat GM1-gangliosidosis by heterotopic insertion of GLB1 in a murine model
335	Alejandra Puentes-Tellez	Perspectives into the spatial organization of HGSNAT protein structure for Sanfilippo syndrome type C (MPS IIIC)
336	Ana Puhl	Combining machine learning and in vitro approaches to identify potential chaperones for lysosomal diseases
337	Marisa Pulcrano	Translating a novel fetal therapy for lysosomal diseases into clinical care: The race for approval to treat one patient with mucopolysaccharidosis type VII
338	Zully Pulido	Recombinant hexosaminidases conjugated to magnetite nanoparticles: Alternative therapeutic treatment routes in GM2 fibroblasts
339	Ruth Pulikottil-Jacob	Relevance and comprehension of patient-reported outcome measures of mobility, physical function, and speech among patients with late-onset GM2 gangliosidoses
340	Alexander Pushkov	Next generation sequencing is useful for the diagnosis of mucopolysaccharidosis type III in Russian patients
342	Uma Ramaswami	Routes to diagnosis of Fabry disease according to patient age and geographic distribution
343	Johana Ramírez Borda	High risk screening for mucopolysaccharidosis type III B in Colombia: Application of a micromethod for the analysis of alpha-N-acetylglucosaminidase in dried blood samples collected on filter paper
344	Sarang Rastogi	Family survey results of 7 children with atypical neuronal ceroid lipofuscinosis type 2 (CLN2) disease in the United States
345	Sarang Rastogi	Results of a family survey on healthcare resource needs in children with neuronal ceroid lipofuscinosis type 2 (CLN2) disease in the US and Canada
346	Shoshana Revel-Vilk	Lyso-Gb1 as a biomarker of treatment outcomes in Gaucher disease: An evaluation of data from the Gaucher Outcome Survey (GOS) registry
347	Kristina Robinson	Tay-Sachs disease carrier screening: Comparative analysis of NGS-based sequencing and enzyme testing results
348	Camille Rochmann	Natural history of late-onset GM2 gangliosidosis: Four years of data collected at NTSAD annual conferences
349	Camille Rochmann	Perspective of patients and caregivers on the disturbance of the disease manifestations of Gaucher disease type 3 on patient's daily lives
350	Tatiane Roncato	“Little people, big world”: Morquio syndrome type A (MPS IVA) in adult life
351	Nilton Rosa Neto	FGF-23 and osteocalcin levels are not associated with cardiovascular manifestations in Fabry disease
352	Nilton Rosa Neto	Hyperferritinemia in Fabry disease: Higher serum TNF-α levels and elevated frequency of hyperlipidemia, chronic renal failure and heart failure
353	Nilton Rosa Neto	Profile of classic Fabry disease patients with history of recurrent fever
354	Nilton Rosa Neto	Access to enzyme replacement therapy and to ancillary tests in Brazilian Fabry disease patients
355	Nilton Rosa Neto	Early retirement in classic Fabry disease patients: Clinical aspects in a Brazilian cohort

356	Nilton Rosa Neto	Kidney transplantation in classic Fabry disease patients: Low diagnostic yield in kidney biopsies before transplantation
357	Nilton Rosa Neto	Chronic back pain in Fabry disease patients: Association with vertebral fractures
358	Nilton Rosa Neto	Deterioration of bone strength as assessed by HR-pQCT in male Fabry disease patients
359	Nilton Rosa Neto	Novel alpha-galactosidase A mutations compatible with classic Fabry disease: Clinical characteristics and previous misdiagnosis
360	Tamanna Roshan Lal	Newborn screen for MPS I - an update of the Washington, DC experience
361	Emory Ryan	Scoliosis in chronic neuronopathic Gaucher disease: Characterization in the treatment era
362	Stephanie Sacharow	Sanfilippo syndrome type A (MPS IIIA) with attenuated genotype manifesting in infancy with dysostosis multiplex
363	Ryo Saito	Development of neuropathological model using Niemann-Pick type C patients derived iPSCs
364	Amir Salahi	Assessment of the diets of Fabry disease patients correlated with reported gastrointestinal symptoms
365	Diana Salazar	Effects of cyclodextrin analogs on psychosine cytotoxicity
366	Richard Sam	Development of a human 3D midbrain organoid model for investigating the link between glucocerebrosidase and Parkinson disease
367	Ana Sanchez	Long-term enzyme replacement therapy for MPS VI: 8-year follow up of a Colombian patient
368	Maria Dolores Sanchez-Niño	Transcriptomics characterization of lyso-Gb3 impact on cultured wild-type human podocytes
369	Emilie Sandfeld	Genomic correction of Pompe disease knock-in mouse myoblasts via CRISPR-Cas9 homology-directed repair
370	Markku Savolainen	Validation of the GED-C point-scoring system in true Gaucher disease patients
371	Kirill Savostyanov	Selective screening for nephropathic cystinosis among high-risk contingents of the children population in Russia
372	Raphael Schiffmann	Lyso−Gb3 is not a predictive biomarker of treatment response in migalastat-treated patients with migalastat-amenable variants
373	Raphael Schiffmann	Venglustat combined with imiglucerase positively affects neurological features and brain connectivity in adults with Gaucher disease type 3
374	Angela Schulz	Cerliponase alfa for the treatment of CLN2 disease in an expanded patient cohort including children younger than three years: Interim results from an ongoing clinical study
375	Angela Schulz	Persistent treatment effect of cerliponase alfa in children with CLN2 disease: A >4 year update from an ongoing multicenter extension study
376	Christoph Schwering	Development of the “Hamburg best practice guidelines for ICV-enzyme replacement therapy (ERT) in CLN2 disease” based on 5 years treatment experience in 48 patients
377	Christine Serratrice	Coagulation parameters in adult patients with type 1 Gaucher disease
378	Christine Serratrice	Natural history of untreated patients with type 1 Gaucher disease
379	Volkan Seyrantepe	Alteration in redox homeostasis in early-onset Tay-Sachs disease mouse model
380	Pankaj Sharma	Analysis of rare variants in lysosomal pathway genes in patients with Gaucher disease with and without Parkinson disease
381	Reena Sharma	Role of 3-dimensional (3D) reconstruction of radiology images and virtual endoscopy in the assessment of airways in adult mucopolysaccharidosis patients
382	Morgan Simmons	Key signs and symptoms associated with GLA variant detection in relatives of an individual with a known GLA variant
383	Paul Solari	Increasing awareness and earlier testing to improve MPS patient outcomes: Simply Test for MPS enzyme-panel program results

384	Rodrigo Starosta	Morphological evidence for hepatic canalicular dysfunction in ERT-treated patients with Gaucher disease
385	Chanan Stauffer	Smoldering myeloma as the initial presentation of Gaucher disease type 1
386	Dean Suhr	What do researchers, clinicians, industry employees, advocacy leaders, 30 million with rare disease, and 69 million of the general public have in common?
387	Dean Suhr	Scientific, clinical, social, and policy status and considerations for implementing newborn screening of metachromatic leukodystrophy
388	Gere Sunder-Plassmann	Baseline patient characteristics of followME, a new, patient-centric, prospective, observational Fabry registry that evaluates migalastat, ERT, and a natural history cohort
389	Xinze Tan	SAAMP 3.0©: computational biology to predict the phenotype of a missense mutation for lysosomal diseases
390	Satowa Tanaka	A novel approach to CNS dysfunction of Pompe disease with a fusion protein consisting of anti-transferrin receptor antibody and GAA enzyme
391	Daisy Tapia	Fabry patients' perspectives on multidisciplinary clinics
392	Sophie Thomas	Supporting adults living with mucopolysaccharide (MPS) diseases: Understanding current experiences and future challenges
393	Sophie Thomas	Prevalence of intestinal disease as terminal event in mucopolysaccharidosis type III - a study of 136 deceased patients
394	Lorraine Thompson	Does the hormonal replacement therapy (HRT) increase the risk of cerebrovascular events in females with Fabry disease?
395	Weihua Tian	Long-acting glyco-design (LAGD) for improved kinetics and distribution of α-galactosidase A
396	Shunji Tomatsu	Newborn screening for mucopolysaccharidoses: Measurement of glycosaminoglycans by LC-MS/MS
397	Shunji Tomatsu	Cochlear implantation in a patient with mucopolysaccharidosis type IVA
398	Shunji Tomatsu	Safety study of sodium pentosan polysulfate for adult patients with mucopolysaccharidosis type II
399	Shunji Tomatsu	Therapeutic options for mucopolysaccharidoses: Current and emerging treatments
400	Shunji Tomatsu	Activity of daily life in patients with mucopolysaccharidosis type II after hematopoietic stem cell transplantation
401	So-Fai Tsang	Mixed safety signals as a result of use of the INN “imiglucerase” for three different products that are not biosimilars: Analysis of adverse events in the Sanofi Genzyme Global Safety database
402	Julie Ullman	Novel FACS based method demonstrates CNS cell-type distribution and efficacy of a BBB penetrant ERT in a mouse model of MPS II
403	Kati Valtola	Family with the rare GLA-Thr410Ala mutation
404	Nato Vashakmadze	Respiratory system involvement in Russian patients with mucopolysaccharidosis: Effects of enzyme replacement therapy
405	Ana Vásquez Salazar	Beta-glucosidase analysis in dried blood collected on filter paper (DBS), report of a new method applied to the population and patients with suspected Gaucher disease (GD)
406	Jesús Villarrubia	Diagnosis of 4 novel cases of adult acid sphyngomielynase deficiency by screening with next-generation sequencing gene panels
407	Birgitte Volck	Gb3 substrate in endothelial cells of renal peritubular capillaries was reduced in a previously untreated classic Fabry male patient treated with AVR-RD-01 investigational lentiviral gene therapy
408	Ashley Volz	Shortening the time between symptom onset and diagnosis for CLN2 disease: results from Behind the Seizure™, a no-cost epilepsy gene panel testing program
409	Eric Wallace	The unmet need in Fabry disease: A retrospective analysis of healthcare claims in the United States reveals significant burden of illness in ERT-treated patients

410	Susanne Walls	Lyso-Gb3: A valid follow-up marker of treatment in male Fabry patients?
411	Ilana Walters	A comparison of the gut microbiome in children affected by Fabry disease and their unaffected siblings: A pilot project
412	Raymond Wang	Intracerebroventricular cerliponase alfa for CLN2 disease: Clinical practice considerations from US clinics
413	Raymond Wang	Long-term safety and efficacy of vestronidase alfa, rhGUS enzyme replacement therapy, in subjects with mucopolysaccharidosis type VII
414	Christoph Wanner	Rationale and design of the MODIFY study: A phase 3 multicenter, double-blind, randomized, placebo-controlled, parallel-group study to determine the efficacy and safety of lucerastat oral monotherapy in adult subjects with Fabry disease
415	David Warnock	Pegunigalsidase alfa, a novel PEGylated ERT, evaluated in Fabry patients with progressing kidney disease, RCT study design
416	Jon Washburn	Reducing false positives in newborn screening for lysosomal disorders
418	Grzegorz Wegrzyn	Underestimated aspect of mucopolysaccharidosis pathogenesis: Global changes in cellular processes revealed by transcriptomic studies and experimental analyses
419	Connie Wehmeyer	A comparison of clinical outcomes and transplant complications/morbidity with early (<4 months of age) versus late (~ 1 yr of age) hematopoietic stem cell transplant in sibling pairs with a diagnosis of MPS I (Hurler syndrome)
420	Ashley Whited	Natural history of white matter development in infantile Krabbe disease
421	Eva Wibbeler	Cerliponase alfa for the treatment of atypical phenotypes of CLN2 disease: A retrospective case series
422	Shanna Widera	Newborn screening for Fabry disease in the state of Illinois has led to the development of a unique clinic called “Fabry Family Clinic”
423	Frits Wijburg	Phase 2-3 gene therapy trial using adeno-associated virus vector for patients with mucopolysaccharidosis type IIIA
424	Jill Wood	Histological characterization of rod-cone retinal degeneration in a mouse model of mucopolysaccharidosis type IIIC
425	Michelle Wood	Correcting dynamic foot abnormalities improves gait and function in mucopolysaccharidosis type III patients
426	Chen Wu	An update on biomarkers of 7-ketocholesterol, lysosphingomyelin, bile acid-408 and glucosylsphingosine for Niemann-Pick disease type C
427	Shirley Wu	Extrapolation of migalastat tissue concentrations in mice as a predictor of human tissue concentrations
428	Hiroyuki Yamakawa	A case of myeloma cast nephropathy complicated by Fabry disease after withdrawal from dialysis
429	Dongshan Yang	A rabbit model of nephropathic cystinosis
430	Isabel Yoon	Long-term neurodevelopmental, neurophysiological, and neuroradiological outcomes of hematopoietic stem cell transplantation for treatment of late-infantile metachromatic leukodystrophy
431	Sarah Young	Characterization of dermatan sulfate and chondroitin sulfate in tissues from a mouse model of mucopolysaccharidosis type VI using UPLC-MS/MS
432	Silvana Zanlungo	ABL1 kinase inhibitors for the treatment of Niemann-Pick disease type A
433	Xiangli Zhao	The therapeutic effect of progranulin derived Pcgin on neuronopathic Gaucher disease
434	N Zhurkova	Russian patients with GM1-gangliosidosis
435	Grace Zimmerman	Methods for quantitative gait analysis in CLN3 disease
436	Ari Zimran	Real life data on the safety and efficacy of ambroxol for patients with Gaucher disease or GBA-related Parkinson disease
LB-01	Carolina Aranda	Enzyme replacement therapy (ERT) with rapid desensitization protocol: Latin America experience
LB-02	Roberto Barriales-Villa	Design of a Fabry disease pedigree project in Spain: Red Fabry, a novel approach

LB-03	Xavier Barril	Allosteric modulators improving biodistribution of recombinant laronidase in MPS1
LB-04	Elfrida Benjamin	Humoral immune responses to ATB200 in the first-in-human study of ATB200/AT2221 in patients with Pompe disease: Results from the phase 1/2 ATB200-02 trial
LB-05	Elizabeth Berry-Kravis	Intrathecal adrabetadex for the treatment of Niemann-Pick disease type C1
LB-06	Daniel Bichet	Migalastat has a low incidence rate of composite clinical outcomes at long-term follow-up in patients with Fabry disease who previously received enzyme replacement therapy
LB-07	Kendra Bjoraker	Individualized education program (IEP) data as endpoint for clinical trials and natural history studies: The 180 Education Outcome Measure
LB-08	Heather Cahan	An observational study investigating potential treatment-responsive CNS biomarkers in subjects with mucopolysaccharidosis type II (MPS II, Hunter Syndrome)
LB-09	Yu-Ting Chiu	Natural product inspired combinatorial chemistry enables us to discover small molecules for the potential treatment of lysosomal disorders
LB-10	Emily de los Reyes	CLN6 Batten disease natural history
LB-11	Maria Duque Lasio	Successful desensitization protocol in female patient with Fabry disease and positive IgE antibody to agalsidase beta
LB-12	Sarah Elsea	Untargeted metabolomic analysis of individuals with MPS II reveals a complex metabolic profile supporting the need for monitoring nutrition and renal and liver function
LB-13	Maria Escolar	RGX-121 gene therapy for severe MPS II (Hunter syndrome): Interim results of an ongoing first in human trial
LB-14	Kaoru Eto	Usefulness of Fabry Stabilization Index in Japanese female patients with Fabry disease
LB-15	Bernhard Gentner	Extensive metabolic correction of mucopolysaccharidosis type I (MPS IH, Hurler syndrome) by hematopoietic stem and progenitor cell (HSPC) based gene therapy (GT): Preliminary results from a phase I/II trial
LB-16	Vykuntaraju Gowda	Study of clinical, biochemical, radiological, molecular and histological profile of neuronal ceroid lipofuscinoses (NCL) from India
LB-17	Nathalie Guffon	Treatment of mucopolysaccharidosis type VI patients with odiparcil alone or in addition to enzyme replacement therapy: A phase IIA study
LB-18	Caroline Hastings	Hydroxypropyl beta cyclodextrin (Trappsol Cyclo) administered intravenously in patients with Niemann-Pick disease type C disease reduces cholesterol in liver tissue
LB-19	Harrison Jones	Determining the diagnostic utility of the identification of tongue involvement in late-onset Pompe disease
LB-20	Dawn Laney	Fabry appointment companion: A novel tool to improve patient and HCPs communication and satisfaction with disease management.
LB-21	Pamela Lavoie	Ultra-performance liquid chromatography-tandem mass spectrometry test for lyso-Gb3 and related analogs in dried blood spots for monitoring and follow up of Fabry disease patients
LB-22	Raffaele Manna	A triple syndromic complex distinguishes Fabry disease patients from patients with autoinflammatory fevers
LB-24	David Moreno-Martinez	Standardising clinical outcomes for Fabry disease clinical trials: A Delphi consensus
LB-25	David Moreno-Martinez	Influence of treatment on stroke recurrence in a large cohort of Fabry disease patients
LB-26	William Mueller	Modified asparagine (GlcNAc-Asn) identified as a biomarker for NGLY1 deficiency.
LB-27	Albina Nowak	Use of agalsidase alfa in the elderly: Clinical outcomes from the Fabry Outcome Survey
LB-28	Shungo Okamoto	Experience of enzyme replacement therapy for mucopolysaccharidosis type VII
LB-29	Charles O'Neill	Neurofilament light is a treatment-responsive biomarker in CLN2 disease
LB-30	Maximiliano Ormazabal	New insights in Gaucher cells models: Characterization of a monocyte edited cells using CRISPR/Cas9 technology
LB-31	Ana Puhl	Comparing routes of administration for an enzyme replacement therapy for infantile onset neuronal ceroid lipofuscinosis (CLN1) disease

LB-32	Matthijs Raaben	Identification of genetic modifiers as therapeutic targets for lysosomal diseases
LB-33	Julien Roeser	Behavioral and neurochemical abnormalities in a pharmacologically induced mouse model of the Gaucher disease
LB-34	Nilton Rosa Neto	Use of intravenous lidocaine and pamidronate as adjuvant therapy to Fabry disease pain crises: Report of two cases
LB-35	Paula Rozenfeld	Pathogenesis of Fabry disease nephropathy: Description of key players in fibrosis
LB-36	Aquilino Sánchez	Reduction of agalsidase beta infusion time in patients with Fabry disease: A case series report and suggested protocol
LB-37	Aquilino Sánchez	Desensitization of agalsidase beta in a patient with Fabry disease and infusion associate adverse reaction: Experience and suggested protocol
LB-38	Jessica Scherr	Comparing developmental outcomes of children with CLN2 disease receiving cerliponase alfa to a natural history cohort
LB-39	Daesung Shin	Brainstem development requires galactosylceramidase and is critical for the pathogenesis of Krabbe disease
LB-40	Seung-Yub Shin	Development of a novel glucosylceramide synthase (GCS) inhibitor with increased blood-brain barrier penetration for treatment of Gaucher disease
LB-41	Shunji Tomatsu	Thermostable keratanase-immobilized column: A device for plasmapheresis therapy for mucopolysaccharidosis type IV
LB-42	Stephanie Trudel	Identification of surrogate cerebrospinal fluid biomarkers for monitoring gene therapy efficiency in Hurler syndrome
LB-43	Osvaldo Uchitel	ASIC1a channels are upregulated by Fabry disease accumulation of sphingolipids
LB-44	Todd Vanyo	The gangliosidoses: Comparisons of GM1-gangliosidosis and GM2-gangliosidosis in 54 patients.
LB-45	Patricia Varela	Early diagnosis of Fabry disease by renal ultrasound imaging
LB-46	Stephan vom Dahl	Attainment of therapeutic goals with ERT in patients with type 1 Gaucher disease in Germany: Interim results from the MUTIG study
LB-47	Jill Weimer	Gene therapy rescues pathological and behavioral deficits in CLN8-Batten disease